

Patient Advocate Leads Drive to Add Rare Diseases to CIRM's Stem Cell Bank

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Oakland, CA – Christina Waters is a rare kind of person: creative, compassionate, and committed to creating change. Now she is using those rare talents to help children battling rare diseases. She is helping add blood and skin samples from these children to be turned into stem cells and used as a resource for researchers to better understand the diseases and hopefully develop new treatments for them.

Waters runs Rare Science a non-profit organization that focuses on children with rare diseases by partnering with patient family communities and foundations. She says of the 7,000 identified rare diseases 50 percent affect children, and 30 percent of those children die before their 5th birthday.

"The biggest gap in drug development is that we are not addressing the specific needs of children, especially those with rare diseases. We need to focus on kids. They are our future. If it takes 14 years and \$2 billion to get FDA approval for a new drug, how is that going to address the urgent need for a solution for the millions of children across the world with a rare disease? That's why we created Rare Science. How do we help kids right now, how do we help the families? How do we make change?"

One of the patient communities she has partnered with is ADCY5.org. ADCY5-related dyskinesia is an abnormal involuntary movement disorder caused by a genetic mutation that results in muscle weakness and severe pain. Because it is so rare, little research has been done on developing a deeper understanding of it, and even less on developing treatments.

That's why Waters has arranged for the collection of blood and skin samples from children with ADCY5 around the world, and submitting them to the CIRM Induced Pluripotent Stem Cell (iPSC) Bank. The samples will be turned into iPS cells that have the ability to be turned into any other type of cell in the body, enabling researchers to study how the disease progresses, and to develop and test new drugs or other therapies.

"Adding samples from rare diseases like ADCY5 is important for both the patients and the researchers," says Jonathan Thomas, PhD, JD, Chair of the CIRM Board. "Just knowing the gene that causes a particular problem is only the beginning. By having the iPSCs of individuals, we can start to investigate the diseases of these kids in the labs. Deciphering the biology of why there are similarities and dissimilarities between these children could the open the door for life changing therapies."

In addition to these samples, the iPSC Bank hopes to ultimately collect around 3,000 samples from people all over California. These will come from healthy people and people with a variety of different diseases, disorders and conditions, including:

- Heart, lung and liver diseases
- Blinding eye diseases
- Childhood neurological disorders, such as epilepsy autism and cerebral palsy
- Alzheimer's disease

The goal is to create the largest iPSC bank in the world with a collection of high quality stem cell lines, developed from thousands of individuals, that can then be made available to researchers throughout California and around the world.

Waters hopes her efforts with ADCY5 will serve as a model for other rare diseases, creating stem cell lines from them to help close the gap between discovery research and clinical impact.

She says she has already been contacted by a researcher in Germany who dropped his research on ADCY5 because it was impossible to get the cells he needed. Now he says, because of these cells, he will be able to start again.

After a career in biotech and pharmaceuticals, Waters says she feels she has found her true purpose, as a patient advocate for children with rare diseases: "What's most important is empowering patient families and helping them accelerate research to the clinical

solutions they so urgently need for their child."

About CIRM

At CIRM, we never forget that we were created by the people of California to accelerate stem cell treatments to patients with unmet medical needs, and act with a sense of urgency to succeed in that mission.

To meet this challenge, our team of highly trained and experienced professionals actively partners with both academia and industry in a hands-on, entrepreneurial environment to fast track the development of today's most promising stem cell technologies.

With \$3 billion in funding and approximately 300 active stem cell programs in our portfolio, CIRM is the world's largest institution dedicated to helping people by bringing the future of cellular medicine closer to reality.

For more information, go to www.cirm.ca.gov

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